



ACT SHEET FOR POSITIVE NEWBORN SCREENING RESULT FOR CONGENITAL ADRENAL HYPERPLASIA

Disease Category: Adrenal endocrinopathy

MEDICAL EMERGENCY: TAKE THE FOLLOWING IMMEDIATE ACTIONS:

- ❖ Contact family **IMMEDIATELY** to inform them of the newborn screening result and ascertain clinical status.
- ❖ Consult with Pediatric endocrinologist, having the following information available (sex, age at NBS sampling, birth weight) and refer, if needed.
- ❖ IMMEDIATELY examine the newborn (ambiguous genitalia or non palpable testes), Assess for salt-wasting symptoms - (emesis, diaphoresis, pallor, lethargy, excessive weight loss relative to birth weight, hyperventilation, dry mucosa).
- ❖ Initial lab work (Do not administer any medications until all specimens have been collected): electrolytes (locally), steroid profile (at reputable laboratory)
- ❖ IV fluid (D10 0.5NS) replacement if dehydrated.
- ❖ Admit patient to hospital for observation due to failed NBS (ICD-9 796.6). Even if minimal or no symptoms, monitoring is critical.
- ❖ Consider administration of IV or IM Solu-Cortef 50mg/M² **after** obtaining steroid profile

Pediatric Endocrinology contacts:

Nebraska Medical Center Pediatric Endocrinology (Omaha)
Kevin Corley, MD
402 559-9197

Children's Hospital Pediatric Endocrinology (Omaha)
Monina Cabrera, MD, or Jean Claude DesMangles MD
402 955-3871

Meaning of the Screening Result:

Congenital Adrenal Hyperplasia (CAH), 21-OH deficiency; stress or immaturity are possible causes.

Condition Description:

Lack of adequate adrenal cortisol and aldosterone, and increased androgen production.

Confirmation of Diagnosis:

Diagnostic tests include serum 17-OHP (increased), serum electrolytes (reduced sodium and increased potassium), and blood glucose (reduced). Additional tests such as specific steroid profiles may be recommended by the specialists.

Clinical Expectations:

Ambiguous genitalia in females. Infant may appear to be male with non-palpable testes. Males will appear normal. Feeding problems, life-threatening adrenal crisis, shock, and **death** in males and females. Finding could also be a false positive associated with stress or prematurity.

Additional Information:

- ❖ New England Metabolic Consortium - www.childrenshospital.org/newenglandconsortium
- ❖ Gene Tests/Gene Clinics - www.genetests.org
- ❖ U.S. National Newborn Screening & Genetics Resource Center - www.genes-r-us.uthscsa.edu
- ❖ Congenital Adrenal Hyperplasia Research and Education Foundation (CARES Foundation) www.caresfoundation.org